



IL23R gene

interleukin 23 receptor

Normal Function

The *IL23R* gene provides instructions for making a protein called the interleukin 23 receptor. This protein is embedded in the cell membrane of several types of immune system cells, including T cells, natural killer (NK) cells, monocytes, and dendritic cells. These cells identify foreign substances and defend the body against infection and disease.

At the cell surface, the interleukin 23 receptor interacts with a protein called interleukin 23. These two proteins fit together like a lock and its key. Interleukin 23 is a cytokine, which is a type of protein that regulates the activity of immune system cells. When interleukin 23 binds to its receptor, it triggers a series of chemical signals inside the cell. These signals promote inflammation and help coordinate the immune system's response to foreign invaders such as bacteria and viruses.

Health Conditions Related to Genetic Changes

ankylosing spondylitis

Several variations (polymorphisms) in the *IL23R* gene have been found to influence the risk of ankylosing spondylitis. One of these variations appears to reduce the likelihood of developing this disorder. This genetic change alters a single protein building block (amino acid) in the interleukin 23 receptor, replacing the amino acid arginine with the amino acid glutamine at protein position 381 (written as Arg381Gln). Other *IL23R* variations appear to increase the risk of developing ankylosing spondylitis. It is not clear how these changes are related to a person's risk of developing this disorder, but studies suggest that the effects of *IL23R* variations are likely related to the interleukin 23 receptor's role in inflammation. Other genetic and environmental factors, many of which are unknown, also affect the chance of developing ankylosing spondylitis.

Crohn disease

Several variations in or near the *IL23R* gene have been found to influence the risk of developing Crohn disease. These associations have been found primarily in white populations. For example, Arg381Gln, which is a protective factor for ankylosing spondylitis, also appears to reduce the risk of developing Crohn disease. Although it is unclear how this change protects against Crohn disease, researchers believe that

the receptor's role in triggering inflammation in the intestinal walls may underlie its connection with this disorder.

psoriatic arthritis

ulcerative colitis

other disorders

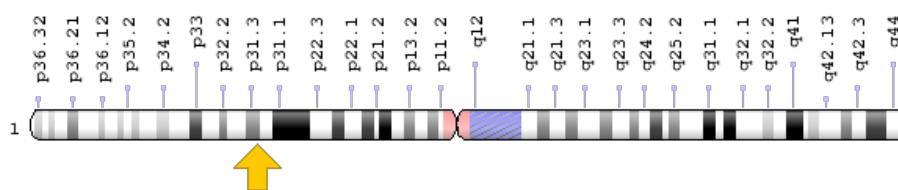
Variations in the *IL23R* gene have also been associated with a skin disorder called psoriasis. People with this chronic inflammatory condition have patches of red, irritated skin that are often covered by flaky white scales. Psoriasis likely results from a malfunction of the immune system in which the body's immune response turns against itself, attacking healthy skin cells by mistake.

Each of the known *IL23R* variations changes a single amino acid in the interleukin 23 receptor. One of these variations, Arg381Gln, appears to reduce the risk of developing psoriasis. (This variation has also been shown to protect against ankylosing spondylitis and Crohn disease, which are other disorders associated with chronic inflammation.) Other *IL23R* variations may increase the risk of developing psoriasis. Researchers suggest that changes in the *IL23R* gene may contribute to general problems with regulation of the immune system, which may help explain why these variations are related to several different disorders characterized by immune system dysfunction.

Chromosomal Location

Cytogenetic Location: 1p31.3, which is the short (p) arm of chromosome 1 at position 31.3

Molecular Location: base pairs 67,138,638 to 67,259,979 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- IL-23R
- IL23R_HUMAN
- interleukin-23 receptor

Additional Information & Resources

Educational Resources

- National Institute of Arthritis and Musculoskeletal and Skin diseases: Psoriasis
https://www.niams.nih.gov/Health_Info/Psoriasis/

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28IL23R%5BTIAB%5D%29+OR+%28interleukin+23+receptor%5BTIAB%5D%29%29+OR+%28IL-23R%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- INTERLEUKIN 23 RECEPTOR
<http://omim.org/entry/607562>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_IL23R.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=IL23R%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=19100
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/149233>
- UniProt
<http://www.uniprot.org/uniprot/Q5VWK5>

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